

LETTER TO THE EDITOR

Indirect inguinal hernia among Bedouins

We read with interest the genetic analysis of 280 families with congenital indirect inguinal hernia (IIH) ascertained in Chandong province in China.¹ The study included 202 families with sporadic cases (72.14%) and 78 families with a positive family history (27.85%). In their study, Gong *et al*¹ concluded that congenital IIH is not compatible with a multifactorial threshold model. However, the observed vertical transmission with a high segregation ratio (0.225) suggested an autosomal dominant model of inheritance with reduced penetrance (0.45). The preferential paternal transmission of the trait illustrated in the same study suggested a parental sex influence with a role for maternal genomic imprinting in the aetiology of the condition.²

Here, we report a Bedouin mother and four of her daughters who were found to have congenital IIH and were surgically managed with herniotomy. The affected daughters had seven unaffected sibs, two males and five females. None of the affected members showed any evidence of collagen diseases, for example, Marfan or Ehlers-Danlos syndromes. Karyotyping of their peripheral blood showed a normal female complement (46, XX). Pelvic sonographic evaluation showed normal Müllerian duct derivatives and their hormonal profile was that of normal females.

In this Bedouin family with only affected females in two successive generations (a mother and four daughters), the possibility is again raised of an autosomal dominant mode of inheritance of the trait, but the situation here is presumably the opposite to the suggested maternal imprinting and preferential paternal transmission observed in the study of Gong *et al*.¹ The identification of exclusively affected females in the present Bedouin family would probably suggest genetic heterogeneity of the condition with possible involvement of other sex influenced genes.

Further follow up of familial IIH among Bedouins, with their characteristically large family size and high inbreeding coefficient,³ will help illustrate the contribution of genetic factors to IIH and the possible role of genomic imprinting in this condition.

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1 Gong Y, Shao C, Sun G, *et al*. Genetic study of indirect inguinal hernia. *J Med Genet* 1994;31: 187-92.

- 2 Hall JG. Genomic imprinting: review and relevance to human diseases. *Am J Hum Genet* 1990;46:857-73.
- 3 Farag TI, Teebi AS. Genetic disorders among Bedouins. In: Teebi AS, Farag TI, eds. *Genetic disorders among Arab populations*. Oxford: Oxford University Press, 1996.

BOOK REVIEWS

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Cystic Fibrosis: Current Topics. Editors J A Dodge, D J H Brock, J H Widdicombe. (Pp 369; £70.00.) UK: John Wiley. 1996. ISBN 0-471-96353-4.

The third volume of *Cystic Fibrosis: Current Topics* well illustrates the long road after a disease gene is isolated, but before major advances in treatment ensue. The contents of this volume address some of the important current research areas and many chapters will undoubtedly provide valuable reviews of individual topics.

The choice of "Cell cultures in CF research" for the opening chapter is logical and potentially useful though regrettably it is far from comprehensive and so of limited value. Subsequent reviews of our current understanding of the molecular basis of CFTR chloride ion channel function and of regulation of CFTR activity are good updates. Next come two chapters that concentrate on CFTR in two different epithelia. The first addresses one of the most exciting areas of new investigation, that is, the regulation of other airway epithelial ion channels by CFTR. Dissection of these interactions may well be crucial to our understanding of the molecular basis of CF lung disease. The second addresses the regulation of CFTR abundance, targeting, and trafficking in the intestinal epithelium.

The genetic aspects of CF are addressed in three chapters: an interesting one on the evolution of CF alleles, a topic that can be investigated with increasing accuracy as the mutation and polymorphism screening data collected by members of the CF genetic analysis consortium expands; next, chapters on the efficacy and desirability of neonatal screening for CF, and on some aspects of the psychological consequences for CF carrier screening. Congenital absence of the vas deferens, which now appears in some cases to be an atypical form of CF, is the next topic to be covered in some detail.

The last basic research chapter, which reviews the molecular biology of mucoidy and

the conversion from non-mucoid to mucoid *Pseudomonas aeruginosa* in the CF lung, leads well into the more clinical sector of the book. This concentrates primarily on aspects of CF lung disease: anti-*Pseudomonas* antibiotics, *Aspergillus* lung disease, the role of DNase and other mucolytics in the treatment of CF lung disease, and finally a chapter on the clinical management of CF patients before and after lung transplantation. A review of CF clinical scoring systems rounds up this useful volume.

ANN HARRIS

Introduction to Veterinary Genetics. F W Nicholas. (Pp 317; £17.99.) Oxford: Oxford University Press. 1996. ISBN 0-19-854292-5.

Hearing so much about Mad Cow Disease at the moment reminds us that animals as well as humans can get sick. This book concentrates on genetic disease in animals and, not surprisingly, almost any form of genetic disorder with which we are familiar in humans is also known in one or other of our domesticated animals. Ehlers-Danlos syndrome has been found in cats, dogs, horses, mink, rabbits, cattle, and sheep, congenital adrenal hyperplasia in rabbits, and Chediak-Higashi syndrome in the killer whale and sheep. Molecular analysis is being used to define the genes and mutations. A deletion in a growth hormone receptor gene produces sex linked dwarfism in broiler chickens. A missense mutation in the gene for a calcium release channel is responsible for the malignant hyperthermia syndrome triggered by mild stress which is suffered by pigs. All this information, and much more besides, is contained in this most excellent book, which, as its name suggests, is basically a genetics textbook for those studying or practising the care or production of domestic animals. It is a most comprehensive, and comprehensible, primer on genetics. It is right up to date, accurate, and clear, and it is a delight for a human geneticist to read because it is so interesting. It covers basic genetics, genes, chromosomes, and molecular biology, before turning to disease: single gene disorders, chromosome aberrations, multifactorial conditions, pharmacogenetics, immunogenetics, and population genetics, all exemplified by descriptions of animal genetic diseases. Then it deals with subjects more directly related to animal husbandry, such as relationship and inbreeding, and single genes important to commercial animal breeding, for example, for producing the greatest proportion of medullated sheep hair fibres, which are best for carpet manufacture, and for enabling the easy sexing of 1 day old chicks.

In this book, the phrase "animal models" takes on a new dimension. The book is highly recommended both for its genetics (veterinarians will be well instructed by it) and for the horizons which it broadens for human geneticists.

MARY J SELLER

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Chromosomes: *ISCN 1995. An International System for Human Cytogenetic Nomenclature*, Mitelman F (ed); S Karger, Basel, 1995

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Enzymes: Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry. New York: Academic Press, 1992. Information also available on URL <http://expasy.hcuge.ch/sprot/enzyme.html>

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